INTRODUCTION

SURF1 (also known as Surfeit 1) encodes the SURF1 protein, which is essential for assembly and maintenance of complex IV of the mitochondrial respiratory chain (also known as cytochrome c oxidase or COX). Loss of function mutations in SURF1 are associated with autosomal recessive SURF1 deficiency, which is the most common cause of complex IV deficient Leigh syndrome (OMIM #256000) and the most common single cause of Leigh syndrome in the UK.

This service is provided in collaboration with Dr Garry Brown, alongside our other mitochondrial disease services, and is NHS Highly Specialised Services (HSS) funded for NHS referrals from England and Scotland.

TESTING

All samples MUST be accompanied by a completed Mitochondrial proforma (click here)

<table>
<thead>
<tr>
<th>Diagnostic:</th>
<th>Clinically affected patients</th>
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<tbody>
<tr>
<td>Carrier or Presymptomatic</td>
<td>Relatives of clinically affected patients</td>
</tr>
<tr>
<td>Prenatal:</td>
<td>At risk of having an affected child</td>
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</tbody>
</table>

REFERRALS

- From Hospital Consultants, mainly Clinical Genetics, Neurology, Paediatrics, Metabolic Medicine
- Prenatal referrals are only accepted from Clinical Genetics and / or Prenatal Diagnosis. They must be discussed with the laboratory and arranged in advance.

STRATEGY

- Sequencing of the coding region of SURF1

TECHNICAL INFORMATION

- Sanger sequencing of the exons 1-9 of SURF1

TARGET REPORTING TIMES

- High priority diagnostic tests: 14-28 calendar days
- Routine diagnostic tests: 56 calendar days
- Carrier/Presymptomatic tests: 14 calendar days
- Prenatal testing (includes maternal contamination check): 3 calendar days

N.B. Details are correct for the date of printing only – last updated 14/07/2016